Case Report

A Rare Neurocutaneous Syndrome and A Rare Association

Hasab Alrasoul S Ali[1], Muaz A M Elsayed[2*]

Among the many causes of ataxia, Ataxia -telangictasia is an autosomal recessive[1], multi system disease affecting the skin, nervous system & immune system. It's prevalence has been estimated at 1 to 2 per 100,000 It is a neurodegenerative disorder[2] in which there is progressive cerebellar ataxia, occulocutaneous telangictasias, chronic sinopulmonary disease & high incidence of malignancy associated with variable humoral & cellular immuno deficiency. The abnormal gene has been mapped to the long arm of chromosome 11 (ATM gene)[3]. Carriers has increased sensitivity to ionizing radiation[4]. Telangicatsias tends to develop between the ages of 3-6 years. It also affects the ears, eye lids immunologic abnormality is selective absence of IgA found in 50-80 % ,IgG may also be affected. The T cells percentage is reduced in total & T Helper (CD4) phenotype, with normal or increased percentages of the Suppressor (CD8).
Hodgkin's, non Hodgkin's lymphoma[5] & leukemia[6] develops in 10 % of cases[6]. Other variable body tumors may occur. These patients may become critically ill and need intense care to prevent serious complications or death[7]. The syndrome is associated with abnormal IgA levels with a higher risk of blood malignancies like lymphoma or leukaemia, patients may have telangiectasia else were in the body specially the upper respiratory tract.
The is a case report about one of this rare neuro- cutaneous syndromes which was reported in Khartoum Sudan. Consent from the parents was taken for the purpose of publication and teaching.

Keywords: Ataxia telangiectasia, Sudan, Rare association.

An 11 year old female from central Sudan was brought to medical advice by her parents because of walking difficulties. On further questioning, she was an out come of a normal pregnancy delivered at home in the presence of a mid wife .The child did not cry immediately but fed immediately .Her mile stones were as follows, she sat at 7/ 12, spoke two words at 1 year, walked & controlled her sphincters at 2 years. She completed the vaccination program for her age .No mental sub normality was noticed but at the age of 6 years she stared to have ataxic gait that deteriorated progressively to frequent falls the year after. Also she had a febrile illness associated with squint & poor hearing. No abnormal behavior or sleep distrurbance noticed.

1. Senior Consultant Neurologist
2. Associate professor of neurology
*Correspondance to: Muaz A M Elsayed
Omdurman Teaching Hospital- Neurology unit
P.O.Box 13513 KHT SD
E.mail: muaz_muaz@hotmail.com

Her parents are 2nd degree cousins. No other family members are known to have similar clinical problem or gait disturbance.
The clinical examination revealed an unwell female child with a length of 121 cm, weight 26Kg and normal head circum. No dysmorphic features were noticed but she could not formulate a sentence. Moderate orofacial dyskinetic movements were noticed. There were obvious telangictasiae in both eyes (figure1).
No telangectasiae were seen in the nose, ears or oral cavity. No lymphadenopathy or petechae. Fundi were normal. Normal other cranial nerves but the 8th was difficult to assess. There was more than three patches of vitiligo noticed 3 years ago mainly on the trunk (figure 2).
This is regarded as a very rare association of this disease[8]. The muscle power was around grade 3 there were depressed reflexes but pin prick & vibration sense were normal. There was ataxia of gait & trunk. Examination of the heart, chest & abdomen was normal.

Figuer (1)**:** Showing telangiectasia of the right eye conjunctiva

Figuer (2): Showing vitiligo patch in the abdomen

Her CBC showed normal blood indices apart from mild anemia & unexplained thrombocytosis (512,000). Brain imaging by MRI showed cerebellar atrophy & dilated ventricles. Her IgA levels in serum were normal. Other tests which are important in such cases but were not affordable for the patient are alpha Feto Protein, CEA antigen, other immunoglobulins like IgG and IgM, MRI Chest for hilar assessment.

Figuer (3): The other patient

The other patient who is 7 year old brother had a normal perinatal, natal & postnatal history. He completed the required vaccination program. Noticed to have delayed walking at 2 years. The mother questions his IQ & got worried about fixed redness of both eyes that is becoming more obvious in the last 2 years. This year he became gradually unsteady on gait & tends to fall. He has normal sleep & appetite but he has enuresis. His length was 114 cm, weighed 18 Kg & normal head circumference. The examination findings were in the form of bilateral eye telangictasiae (figuer 3), there was starting vitiligo on the cheek cerebellar ataxia. Other systems examination was unremarkable. His blood tests & MRI brain showed the same findings as his sister.

REFERENCES:

1. Ledezma et al Gac Med Mex. Ataxia telangiectasia. Diagnosis and follow-up in 4 cases. 2013 Jul-Aug;149(4):448-53.
2. Verhagen MM et al Neuropathology in classical and variant ataxia-telangiectasia Neuropathology. 2012 Jun;32(3):234-44.
3. Huh HJ et al Identification of ATM mutations in Korean siblings with ataxia-telangiectasia. Ann Lab Med. 2013 May;33(3):217-20.
4. Byrd PJ et al, Severe reaction to radiotherapy for breast cancer as the presenting feature of ataxia telangiectasia.Br J Cancer. 2012 Jan 17;106(2):262-8.
5. Danby CS et al . Non-hodgkin B-cell lymphoma of the ovary in a child with Ataxia-telangiectasia.J Pediatr Adolesc Gynecol. 2013 Apr;26(2):e43-5.
6. Bielorai B Acute lymphoblastic leukemia in early childhood as the presenting sign of ataxia-telangiectasia variant Pediatr Hematol Oncol. 2013 Sep;30(6):574-82.
7. Lockman JL et al. The critically ill patient with ataxia-telangiectasia: a case series.Pediatr Crit Care Med. 2012 Mar;13(2):e84-90.
8. Sari A[et] al Uncommon associations with ataxia-telangiectasia: vitiligo and optic disc drusen. Ophthalmic Genet. 2009 Mar;30(1):19-22.